Locus 144 (rs288326)

SNiPAcard

Block annotations

Block info

genomic range	chr2:183,702,964-183,703,336
block size	373 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.149 [0.053 – 2.246]	gene(s) hit or close-by	FRZB
phastCons	μ = 0.500 [0 – 1]	eQTL gene(s)	–
GERP++	μ = 0.860 [-1.46 – 3.18]	potentially regulated gene(s)	–
CADD score	μ = 18.154 [4.308 – 32]	disease gene(s)	FRZB

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Osteoarthritis susceptibility 1	risk factor	ClinVar	RCV000005531.1	1
?	HGMD curated	HGMD	CM0910593	1
OSTEOARTHRITIS SUSCEPTIBILITY 1	OMIM curated	OMIM	MIM:605083	1

Disease gene annotation

gene	trait	source DB	source entry/link
FRZB	OSTEOARTHRITIS SUSCEPTIBILITY 1	OMIM	MIM:165720

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FRZB	missense variant	ENST00000295113	NM_001463.3	ENSP00000295113	W/R	Tgg/Cgg	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FRZB	ENST00000295113	NM_001463.3	ENSP00000295113	1